

What will we know when we will have the complete sequence of each of our genomes?

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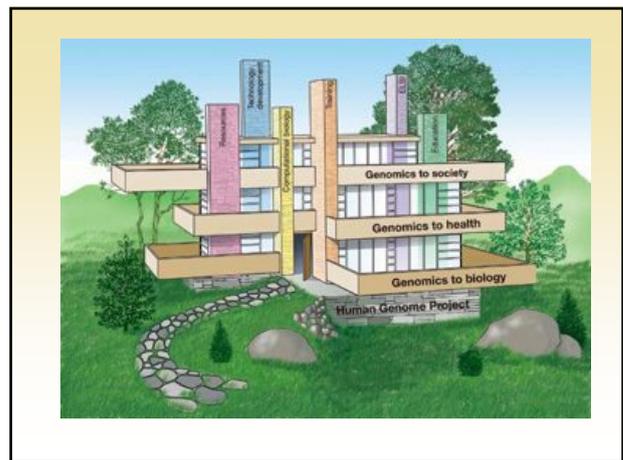
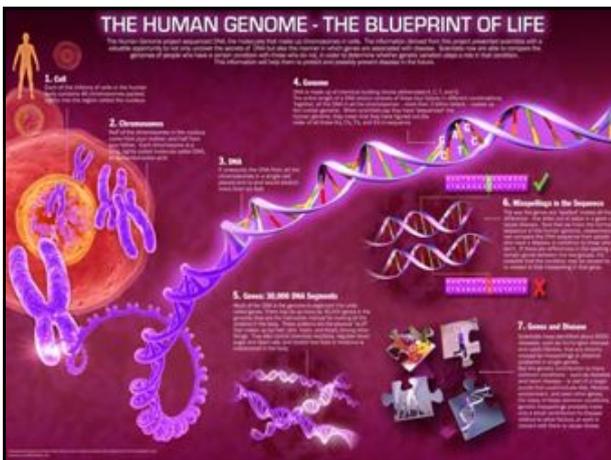
The revolution in the Life Sciences

Living organisms may be viewed as the only part of the natural world whose members contain internal description of themselves.



This is why the whole biology must be rooted in the DNA, and our task is still to discover how these DNA sequences arose in evolution and how they are interpreted in to build the diversity of the living world, including disease.

Sydney Brenner, dec 2012, Science



Forms of genomic variation

Sequence

- **Single base-pair changes** – point mutations (**SNPs**, Single Nucleotide Polymorphisms: 3Mb diff between any two genomes)
- **Small insertions/deletions**
- **Variable number of tandem repeats** (microsatellite, minisatellite)
- **Mobile elements**—retroelement insertions (300bp -10 kb in size)
- **Large-scale genomic variation** (>10 kb)
 - Large-scale Deletions and Amplifications
 - Segmental Duplications / Copy Number Variation (**CNVs**, Up to 15Mb diff. between any two genomes)
- **Chromosomal variation**—translocations, inversions, fusions.

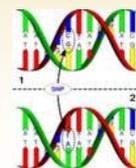
Cytogenetics

Forms of genomic variation

SNPs in the Human Genome

- Normally biallelic (two variants)
- Allelic frequencies present differences between populations.
- Phenotypic effect: normally without effect

- Public databases for SNPs
<http://www.nhgri.nih.gov>,
<http://www.snp.cshl.org>



Linking genes and phenotypes

At the interspecific level



Linking genes and phenotypes

At the intraspecific level



Linking genomic and phenotypic variation

GENOTYPE VARIATION

Genetic Epidemiology,
Statistical Genomics,
Systems Biology...

PHENOTYPE (Disease) VARIATION

Disease as a phenotype

Types of disease:

Molecular classification of human diseases

Exogenous diseases: Infections, Intoxication, Nutritional
(NURTURE)

Genetic diseases:

- Cell? Germinal or somatic
- Alteration?
 - Genic (mutations, ins/del)
 - Chromosomal
- Genome?
 - Nuclear
 - Mitochondrial

Simple pattern of inheritance (NATURE)

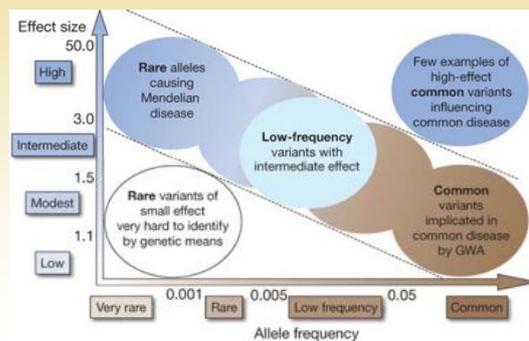
Complex or multifactorial diseases: (NATURE vs NURTURE)

Disease as a phenotype

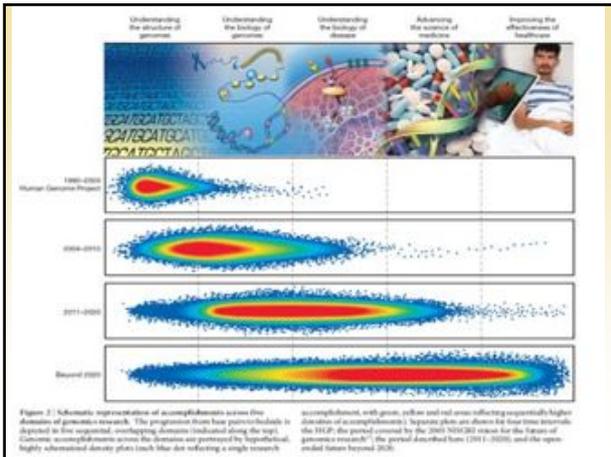
✓ Complex diseases

- High prevalence
 - Cardiopathy
 - Rheumatoid Arthritis
 - Autoimmune disease
 - Psychiatric diseases
 - Cancer
- Relevant genetic component
- No simple Mendelian inheritance pattern
- Several genes with small contribution (susceptibility)
- Multiple alleles interacting among themselves and with the environment.

Allele frequency and genetic effect



From: TA Manolio et al. *Nature* 461, 747-753 (2009) doi:10.1038/nature08490



Sequencing of individual genomes

Genome	Journal/Source	Year
J. Craig Venter	PLoS Biology	2007
James D. Watson	Nature	2008
Marjolein Kriek	LUMC	2008
Yoruban African	Nature	2008
Han Chinese	Nature	2008
Stephen Quake	Nature Biotechnology	2009
George Church	Personal Genome Project	2009
Hermann Hauser	Illumina	2009
2 Koreans	Nature/Genome Research	2009
9 individuals	Knome	2009
Desmond Tutu	Nature	2010
!Gubi	Nature	2010
186 individuals	1000 Genomes Project	2010

Vol 463 | 8 February 2010 | doi:10.1038/nature08795

nature

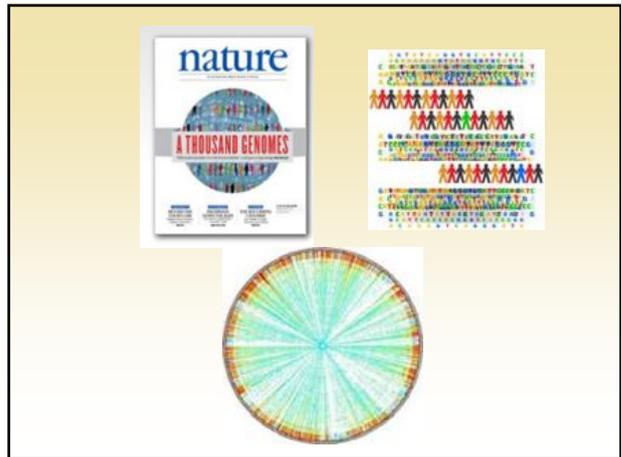
LETTERS

Complete Khoisan and Bantu genomes from southern Africa

Stephan C. Schuster^{1*}, Webb Miller^{1*}, Aakrosh Ratan¹, Lynn P. Tomsho¹, Belinda Giardine¹, Lindsay R. Kasson¹, Robert S. Harris¹, Desiree C. Petersen¹, Fangqing Zhao¹, Ji Qi¹, Can Aikan¹, Jeffrey M. Kidd¹, Yazhou Sun¹, Daniela I. Drautz¹, Pascal Bouffard¹, Donna M. Muzny², Jeffrey G. Reid³, Lynne V. Nazareth³, Qingyu Wang⁴, Richard Burhans⁵, Cathy Riemer⁶, Nicola E. Wittekindt⁶, Priya Moorjani⁷, Elizabeth A. Tindall^{2,7}, Charles G. Danko⁸, Wee Siang Teo^{2,7}, Anne M. Buboltz⁹, Zhenhai Zhang⁹, Qianyi Ma⁹, Arno Oosthuysen⁹, Abraham W. Steenkamp¹⁰, Hermann Oosthuisen¹¹, Philippus Venter¹², John Gajewski¹, Yu Zhang¹, B. Franklin Pugh¹, Kateryna D. Makova¹³, Anton Nekrutenko¹⁴, Elaine R. Mardis¹⁵, Nick Patterson¹⁶, Tom H. Pringle^{1,5}, Francesca Chiaromonte¹⁷, James C. Mullikin¹⁸, Evan E. Eichler¹⁹, Ross C. Hardison¹, Richard A. Gibbs¹, Timothy T. Harkins²⁰ & Vanessa M. Hayes^{2,21*}

Authors (48 in total)
Sequencing data generation (11)
Data analysis (36)

Key words: Khoisan, Bantu, genome, southern Africa, sequencing data generation, data analysis.



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Deep resequencing in human populations

Resequencing of 200 human exomes identifies an excess of low-frequency non-synonymous coding variants [Li et al. 2010](#)

Deep resequencing reveals excess rare recent variants consistent with explosive population growth [Coventry et al. 2010](#)

Demographic history and rare allele sharing among human populations [Gravel et al. 2011](#)

Recent Explosive Human Population Growth Has Resulted in an Excess of Rare Genetic Variants [Keinan et al. 2012](#)

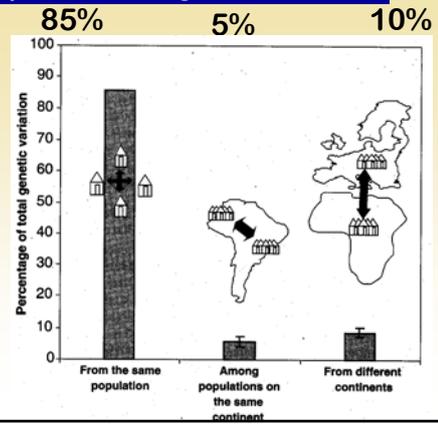
Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes [Tennesen et al. 2012](#)

An Abundance of Rare Functional Variants in 202 Drug Target Genes Sequenced in 14,002 People [Nelson et al. 2012](#)

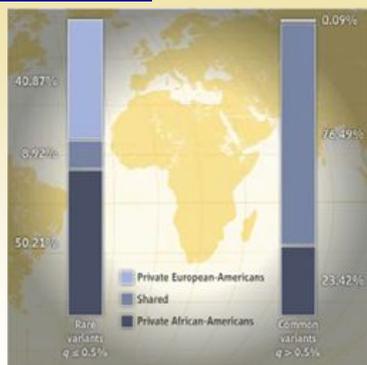
rare is meaningful

human populations show an excess of rare variants, which are enriched for disease causing mutations

The old picture of human genetic variation ...



... and the new picture



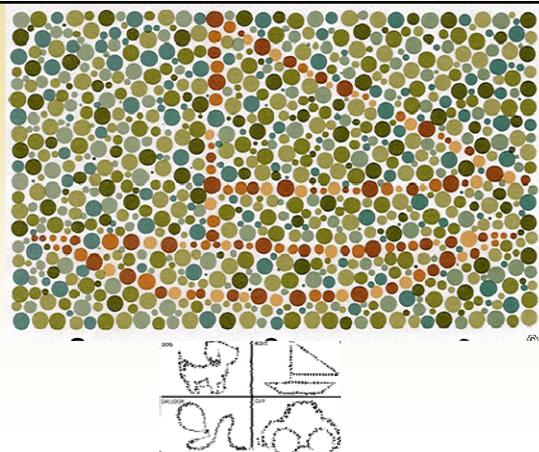
From Casals and Bertranpetti. Science 2012. Human Genetic Variation, Shared and Private.

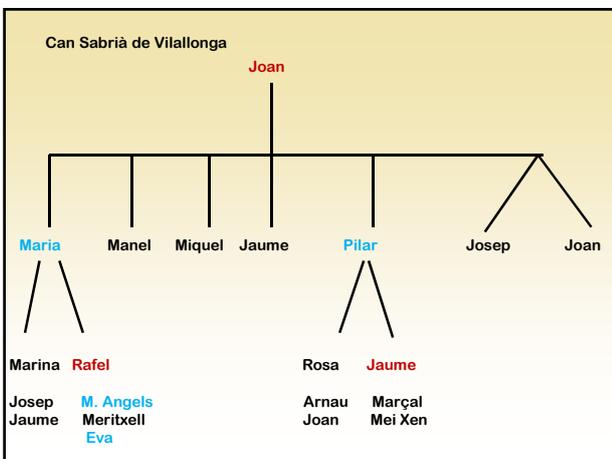
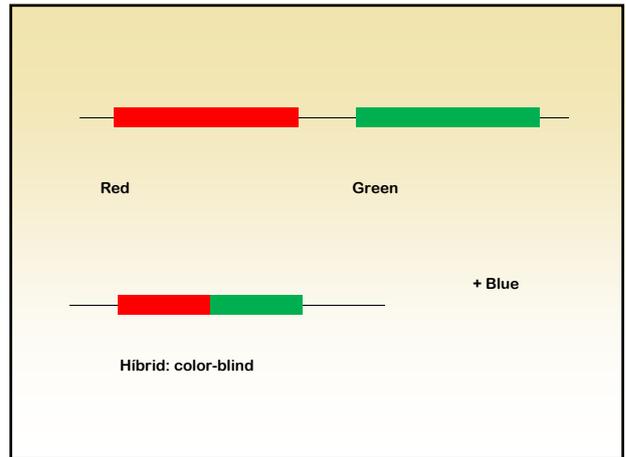
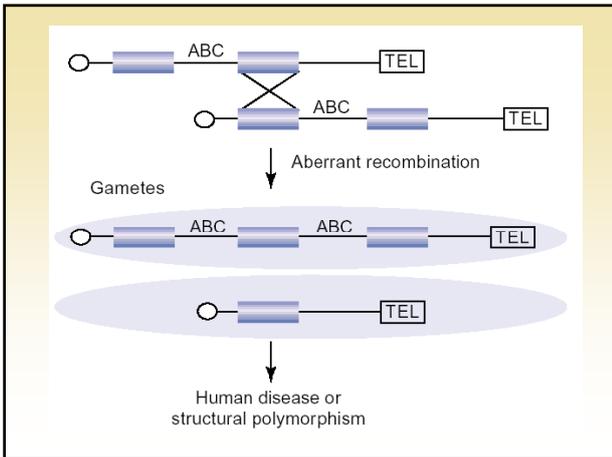
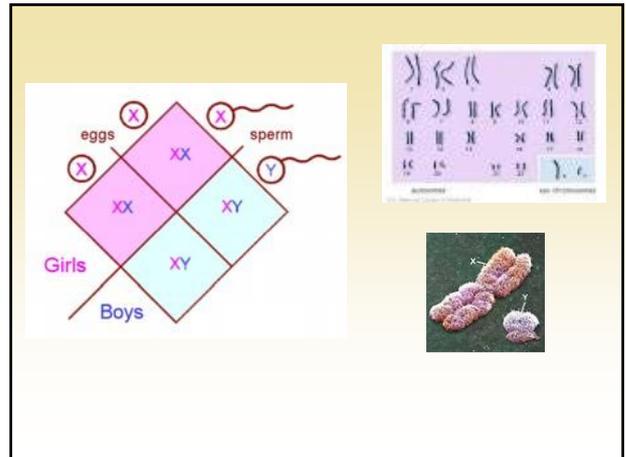
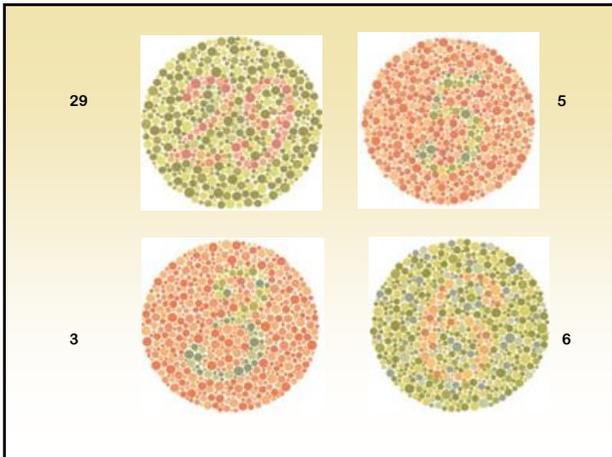
The case of my individual genome

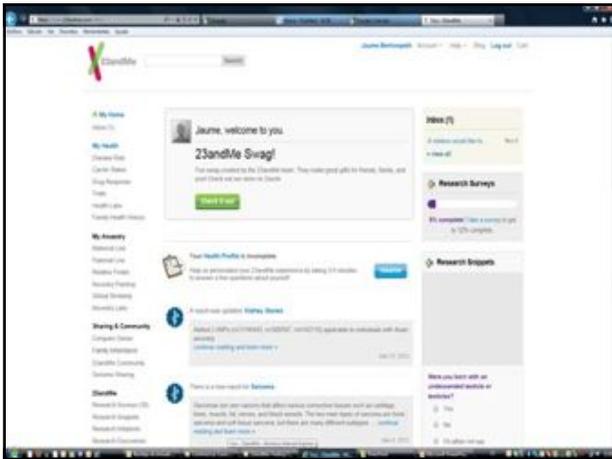
- 1.- Color-blindness
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23andMe Discoveries were made possible by 23andMe members who took surveys.

Show results for Jaume Bertranpeti

See new and recently updated reports +

Disease Risks (118)

Elevated Risks

Disease	Your Risk	Average Risk
Venous Thromboembolism	39.8%	12.3%
Gabapentin	11.1%	7.0%
Rheumatoid Arthritis	3.4%	2.4%
Primary Biliary Cholangitis	0.10%	0.08%
Scleroderma (Limited Cutaneous Type)	0.08%	0.07%

See all 118 risk reports...

Carrier Status (44, 1 locked report)

Variant Present

- Hemochromatosis
- Alpha-1 Antitrypsin Deficiency
- Agmatinase of the Corpus Callosum with Peripheral Neuropathy (ACOPN)
- Autosomal Recessive Polycystic Kidney Disease
- ARSACS
- Bloom's Syndrome
- Ceroid Lipofuscinosis
- Congenital Disorder of Glycosylation Type 1a (PMO-CGG)

See all 44 carrier status...

Traits (55)

Alcohol Flush Reaction	Dem Not Flush
Bitter Taste Perception	Unable to Taste
Earwax Type	Wet
Eye Color	Likely Brown
Hair Curl	Slightly Curlier Hair on Average

See all 55 traits...

Drug Response (20)

Atorvastatin Hypersensitivity	Increased
Clopidogrel (Plavix) Efficacy	Reduced
Warfarin (Coumadin) Sensitivity	Increased
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	Typical
Fluorouracil Toxicity	Typical

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Elevated Risk

Name	Confidence	Your Risk	Avg. Risk	Compared to Average
Venous Thromboembolism	★★★★	39.8%	12.3%	3.22x
Gabapentin	★★★★	11.1%	7.0%	1.55x
Rheumatoid Arthritis	★★★★	3.4%	2.4%	1.42x
Primary Biliary Cholangitis	★★★★	0.10%	0.08%	1.25x
Scleroderma (Limited Cutaneous Type)	★★★★	0.08%	0.07%	1.24x
Alcohol Dependence	★★★			
Alopecia Areata	★★★			
Basal Cell Carcinoma	★★★			
Celiac Disease: Preliminary Research	★★★			

Venous Thromboembolism

23andMe Established Research Report

Jaume Bertranpeti - March 26, 2012

Intended for research and educational purposes. Not for diagnostic use.

Your Genetic Data

You: Increased risk, 39.8 out of 100 men of European ethnicity who share this genotype will get Venous Thromboembolism between the ages of 0 and 79.

Average: The average for this population is 12.3 out of 100.

Technical Report:

Gene or Region	SNP	Genotype	Adjusted Odds Ratio
F5	rs6025	CT	4.69
F2	G002432	GG	1.0

For a list of references for each variant, please see the freely available 23andMe Venous Thromboembolism Report: <https://www.23andme.com/health/venousthromboembolism/>

disease risk

Venous Thromboembolism

4 million live with

Share

Your Data | **How It Works** | Timeline | MD's Perspective | Resources | Technical Report | Community (24)

How the Biology Works

Time—and Clotting—Heals All Wounds

If you are like most people, you've had your share of cuts and scrapes throughout your life. Each time you're injured, whether it be from a tumble off your first bike or a shaving nick caused by a dull blade, you do more than suffer a little pain. The injury also sets off a cascade of biochemical events that ultimately leads to the formation of a blood clot.

When an injury breaks a blood vessel, tissues that normally lie underneath are exposed to the blood stream. Proteins in these tissues interact with cells and proteins in the blood to start the clotting process, which prevents you from bleeding to death.

One group of blood proteins involved in sealing up breaches in blood vessel walls includes the "clotting factors," which act in an organized pathway. After an injury, the first clotting factor is activated. This leads to activation of the second clotting factor in the pathway, which activates the third, and so on. One of the final steps in the pathway is when a protein called "prothrombin" is turned into its active form "thrombin."

1 of 3. The ability of blood to clot prevents it from flowing out of your body uncontrollably in the event of an injury.

Four Genetic Data

Show information for **Jaume Bertranpetit** assuming **European** ethnicity and an age range of **0-75**

Jaume Bertranpetit
39.8 out of 100
Men of European ethnicity who share Jaume Bertranpetit's genotype will develop Venous Thromboembolism between the ages of 0 and 75.

Average
12.3 out of 100
Men of European ethnicity will develop Venous Thromboembolism between the ages of 0 and 75.

What does the Odds Calculator show me?
Use the ethnicity and age range selectors above to see the estimated incidence of Venous Thromboembolism due to genetics for men with **Jaume Bertranpetit's** genotype. The 23andMe Odds Calculator assumes that a person is free of the condition at the lower age in the range. You can use the name selector above to see the estimated incidence of Venous Thromboembolism for the genotypes of other people in your account.

Genes vs. Environment
The heritability of venous thromboembolism is estimated to be 55%. This means that genetics (including unknown factors and known ones such as the SNPs we describe here) and environment play nearly equal roles in this condition. There are a number of environmental factors of various strengths that contribute to venous thromboembolism. Strong risk factors include hip or leg fractures, hip or knee replacement, major surgery or trauma, and spinal cord injury or surgery. Moderate risk factors include antihypertensive, knee surgery, having central venous lines, congestive heart or respiratory failure, hormone replacement or oral contraceptive use, cancer, pregnancy, analytic stroke, previous venous thromboembolism, and thrombophilia. Weak risk factors include bed rest for more than three days, immobility due to sitting (such as a long car or plane trip), specific types of chemotherapy, increasing age, tobacco/cocaine/supercold, obesity, and estrogenic drugs (estrogen).

Decreased Risk

Name	Confidence	Your Risk	Avg. Risk	Compared to Average
Type 2 Diabetes	★★★★	16.7%	25.7%	0.65x ↓
Alzheimer's Disease	★★★★	4.9%	7.2%	0.68x ↓
Age-related Macular Degeneration	★★★★	4.6%	6.9%	0.66x ↓
Rheless Legs Syndrome	★★★★	0.9%	2.0%	0.44x ↓
Melanoma	★★★★	0.7%	2.9%	0.26x ↓
Type 1 Diabetes	★★★★	0.7%	1.0%	0.65x ↓
Esophageal Squamous Cell Carcinoma (ESCC)	★★★★	0.3%	0.4%	0.80x ↓
Multiple Sclerosis	★★★★	0.2%	0.3%	0.69x ↓
Osteoarthritis	★★★★	0.2%	0.5%	0.44x ↓
Stomach Cancer (Gastric Cardia Adenocarcinoma)	★★★★	0.2%	0.2%	0.77x ↓
Exfoliation Glaucoma	★★★★	0.2%	0.7%	0.22x ↓
Celiac Disease	★★★★	0.04%	0.12%	0.33x ↓
Abdominal Aortic Aneurysm	★★★			↓
Atrial Fibrillation: Preliminary Research	★★★			↓
Behçet's Disease	★★★			↓

Typical Risk

Name	Confidence	Your Risk	Avg. Risk	Compared to Average
Obesity	★★★★	65.9%	63.9%	1.03x
Coronary Heart Disease	★★★★	46.7%	46.8%	1.00x
Abetal Fibrinolysis	★★★★	23.0%	27.2%	0.85x
Prostate Cancer	★★★★	13.7%	17.0%	1.01x
Pneumonia	★★★★	9.9%	11.4%	0.87x
Lung Cancer	★★★★	6.9%	8.5%	0.82x
Colorectal Cancer	★★★★	5.0%	5.6%	0.89x
Chronic Kidney Disease	★★★★	3.7%	3.4%	1.06x
Parkinson's Disease	★★★★	1.4%	1.9%	0.85x
Ulcerative Colitis	★★★★	0.7%	0.8%	0.94x
Bipolar Disorder	★★★★	0.10%	0.10%	0.94x
Breast Cancer	★★★★	0.00%	0.00%	1.00x
Lupus (Systemic Lupus Erythematosus)	★★★★	0.00%	0.00%	1.00x
Ankylosing Spondylitis	★★★			↓
Asthma	★★★			↓
Atopic Dermatitis	★★★			↓
Brain Aneurysm	★★★			↓
Coronary Heart Disease: Preliminary Research	★★★			↓
Dupuytren's Disease	★★★			↓

The case of my individual genome

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Carrier Status

Show results for **Jaume Bertranpetit**

23andMe Discoveries were made possible by 23andMe members who took surveys

Locked Reports

Name	Confidence	Confidence
BRCA Cancer Mutations (Selected)	★★★★	
Hemochromatosis	★★★★	Variant Present
Alpha-1 Antitrypsin Deficiency	★★★★	Variant Absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	★★★★	Variant Absent
Autosomal Recessive Polycystic Kidney Disease	★★★★	Variant Absent
ARSA	★★★★	Variant Absent
Bloom's Syndrome	★★★★	Variant Absent
Canavan Disease	★★★★	Variant Absent
Congenital Disorder of Glycosylation Type Ia (FMQ-COG)	★★★★	Variant Absent
Cystic Fibrosis	★★★★	Variant Absent
D-Bifunctional Protein Deficiency	★★★★	Variant Absent
Dihydropyrimidine Dehydrogenase Deficiency	★★★★	Variant Absent
DPD Deficiency	★★★★	Variant Absent
Fenilalanin Dysmetabolism	★★★★	Variant Absent

Chromosome	★★★★	Variant Absent
Gaucher Disease	★★★★	Variant Absent
GRACILE Syndrome	★★★★	Variant Absent
Glycogen Storage Disease Type Ia	★★★★	Variant Absent
Glycogen Storage Disease Type Ib	★★★★	Variant Absent
Primary Hyperparathyroidism Type 2 (PH2)	★★★★	Variant Absent
Hypertrophic Cardiomyopathy (MYBPC3 25bp-deletion)	★★★★	Variant Absent
LARGE-related Junctional Epidermolysis Bullosa	★★★★	Variant Absent
Limb-girdle Muscular Dystrophy	★★★★	Variant Absent
Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	★★★★	Variant Absent
Maple Syrup Urine Disease Type 1B	★★★★	Variant Absent
Mandibuloacral Dysplasia	★★★★	Variant Absent
Neuronal Ceroid Lipofuscinosis (CLN5-related)	★★★★	Variant Absent
Neuronal Ceroid Lipofuscinosis (PPT1-related)	★★★★	Variant Absent
Neuronal Picket Disease Type A	★★★★	Variant Absent
Nyctemeral Breathing Syndrome	★★★★	Variant Absent
Connors 26 Related Sensorineural Hearing Loss	★★★★	Variant Absent
Pandey Syndrome	★★★★	Variant Absent
Thymidylase	★★★★	Variant Absent
Biomineral Chondrodysplasia Punctata Type 1 (BCDP1)	★★★★	Variant Absent
Salla Disease	★★★★	Variant Absent
Skeletal Cell Anemia & Malaria Resistance	★★★★	Variant Absent
Tay-Sachs Disease	★★★★	Variant Absent
Tourette Syndrome	★★★★	Variant Absent
Tuberosin Synthesis Deficiency	★★★★	Variant Absent

carrier status

Hemochromatosis

9 others like this

Share

Your Data | **How It Works** | Resources | Technical Report | Community (32)

How the Biology Works

Introduction

Iron is an essential mineral for the formation of hemoglobin, the oxygen-carrying protein in red blood cells. It is also important for proper brain function, a strong immune system and healthy muscles.

Too much iron in the body, however, can be a problem. Excess iron ends up being stored in the tissues of major organs, especially the liver, heart and pancreas. Over time, "iron overload" can severely damage organs, leading to organ failure and chronic diseases.

There is no way for the body to expel excess iron if the metal's concentration in tissues gets too high. The body can only regulate the amount of iron that is absorbed from the diet. To do this, the body adjusts the number of iron-transporting proteins that are expressed on the surface of cells in the small intestine. If the concentration of iron starts to creep up, the number of transport proteins is decreased. If iron levels are too low, the number of iron transport proteins is increased. Mutations that interrupt this system can lead to an iron overload condition known as hereditary hemochromatosis (HH).



1 of 3 Dietary iron is found in both meat and vegetables, but the body more readily absorbs the iron in meat.

carrier status

BRCA Cancer Mutations (Selected)

6 others like this

Next

Health and Tests: BRCA C...

Your Data | How It Works | Resources | **Technical Report** | Community (32)

Technical Report

Raw genotypes for **Jaume Bertranpetit** in the BRCA1 and BRCA2 genes.

Z3andlife Name	Other Name(s)	DNA Change	Genotype	Result
#000377	135delAG	AG to ---	AG,AG	No copies of the three early-onset breast and ovarian cancer mutations identifiable by Z3andlife. May still carry a different mutation in BRCA1 or BRCA2.
#000378	5382insC	--- to C	---	
#000379	8174delT	T to ---	T,T	

BRCA Cancer Mutations (Selected) and Your Genes

The majority of the 200,000 breast cancer cases diagnosed each year in the United States are "sporadic", meaning the patient has no blood relatives affected with the disease. Only five to 10 percent of breast cancers occur in women with a genetic predisposition for the disease.

BRCA1 and BRCA2 mutations account for most (though not all) cases of inherited breast cancer in women. These mutations are also associated with an increased risk for ovarian cancer. In men, BRCA mutations increase the risk for breast cancer and may also increase prostate cancer risk. Because BRCA1 and BRCA2 encode proteins involved in repairing DNA damage, a process that goes on in virtually all

- The case of my individual genome
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Five genes that shape the human face are identified



Forensic test can predict hair and eye colour from DNA



The test can predict both hair and eye colour from samples left at a crime scene

Scientists have developed a forensic test that can predict both the hair and eye colour of a possible suspect using DNA left at a crime scene.

The test that developed for the test says it could provide reliable tests to solve crime perpetrators cannot be identified through DNA profiling.

traits

Share my health results with family and friends

Show results for **Jaume Bertranpetit** [See new and recently updated reports](#)

23andMe Discoveries were made possible by 23andMe members who took surveys.

Name	Confidence	Outcome
Alcohol Flush Reaction	★★★★	Does Not Flush
Bitter Taste Perception	★★★★	Unable to Taste
Earlobe Type	★★★★	Wet
Eye Color	★★★★	Likely Brown
Hair Curl	★★★★	Slightly Curlier Hair on Average
Lactose Intolerance	★★★★	Likely Intolerant
Malaria Resistance (Duffy Antigen)	★★★★	Not Resistant
Male Pyloric Bacteraemia	★★★★	Increased Odds
Muscle Performance	★★★★	Likely Sprinter
Non-ABO Blood Group	★★★★	Best Report
Nerveous Personality	★★★★	Not Resistant
Resistance to HIV/AIDS	★★★★	Not Resistant
Smoking Behavior	★★★★	Typical
Adiponectin Levels	★★★	Best Report
Appetite Metabolic Detection	★★★	Typical Odds of Detecting
Body Weight	★★★	Best Report
Blood Glucose	★★★	3.12 normal, on Average

ABO Blood Type

By: Nicholas Eriksson

Find out your probable blood type.

[Discuss this feature](#)

[Send feedback](#)

There actually are more than 25 different blood groups that go into determining your particular "type," but you're probably most familiar with the blood groups determined by the ABO and RHD genes. These are the genes that determines whether you will be type O, A, B, or AB, and positive or negative (the Rhesus or Rh factor).

Show probable blood type for: **Jaume Bertranpetit** [Submit](#)

This estimate should not be used for medical purposes

Jaume Bertranpetit's probable ABO blood type:

Type A (A/A)

Jaume Bertranpetit's probable Rh factor:

Positive (+/-)

How is your blood type related to genetics?

The different versions of the ABO gene fall into three main families: A, B and O. Everyone inherits two copies of the ABO gene - one from each parent. This makes six combinations possible: A/O, A/A, B/O, B/B, A/B and O/O. Type A blood results from both the A/O and A/A combinations. Type B blood is caused by the B/O and B/B combinations. To have type O blood, a person must have two "O" versions of the ABO gene. This

The case of my individual genome

- 1.- Color-blindness
- 2.- Disease risk (complex diseases)
- 3.- Carrier status (severe genetic diseases)
- 4.- Traits (physical characteristics)
- 5.- Drug response
- 6.- Ancestry (origins)
 - Maternal origin (mtDNA)
 - Paternal origin (Y-chromosome)
 - Autosomal genome

23andMe Discoveries were made possible by 23andMe members who took surveys.

Name	Confidence	Status
Abacavir Hypersensitivity	★★★★	Increased
Clodrogrel (Flavell) Efficacy	★★★★	Reduced
Warfarin (Coumadin®) Sensitivity	★★★★	Increased
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	★★★★	Typical
Fluorouracil Toxicity	★★★★	Typical
Response to Hepatitis C Treatment	★★★★	Typical
Pseudocholinesterase Deficiency	★★★★	Typical
Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism	★★★★	Not Applicable
Caffeine Metabolism	★★★	Fast Metabolizer
Hepatitis C Treatment Side Effects	★★★	See Report
Antibiotic Response	★★★	Typical Odds of Positive Response
Antidepressant Response	★★	See Report

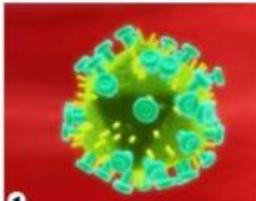
drug response

Abacavir Hypersensitivity

Only a medical professional can determine whether abacavir is the right medication for a particular patient. The information contained in this report should not be used to independently establish an abacavir regimen, or abolish or adjust an existing course of treatment.

Abacavir (trade name Ziagen®; also a component of Epivon® and Trizivir®) is a drug used to treat HIV that works by inhibiting an enzyme the virus needs in order to reproduce. The drug is highly effective, but up to 8% of those treated with abacavir experience a hypersensitivity reaction characterized by fever, rash, respiratory problems, fatigue and nausea. Symptoms usually go away after the drug is discontinued. Everyone who is hypersensitive to abacavir has a particular immune marker called HLA-B*57:01, although people lacking this marker can have other types of adverse reactions. It is now standard practice to test for HLA-B*57:01 before prescribing abacavir.

The following results are based on ★★★★★ Established Research for 1 reported marker



Who	What it Means	Genes vs. Environment
Jaume Bertranpetit	Most likely has at least one copy of HLA-B*57:01 (84% chance). Greatly increased risk of abacavir hypersensitivity.	23andMe reports data for a SNP that predicts the presence of the HLA-B*57:01 marker in the vast majority of people with European ancestry. This SNP is a somewhat less reliable proxy for this marker in people from other ethnic groups. In addition to the data reported here, a person's sensitivity to abacavir may be influenced by clinical and genetic information not presented in this report. Only a medical professional can determine whether abacavir is the right medication for a particular patient. The information contained in this report should not be used to independently establish an abacavir regimen, or abolish or adjust an existing course of treatment.

Abacavir Hypersensitivity and Your Genes

Abacavir is a drug used to treat HIV that works by inhibiting an enzyme the virus needs in order to reproduce. It's highly effective, but up to 8% of those treated with abacavir experience a hypersensitivity reaction.

Everyone who is hypersensitive to abacavir has a particular immune marker called HLA-B*57:01, although people lacking this marker can have other types of adverse reactions. It is now standard practice to test for HLA-B*57:01 before prescribing abacavir.

Citations

Haber et al. (2006). "HLA-B*57:01 screening for hypersensitivity to abacavir." *N Engl J Med* 355(2):260-70.

Martin et al. (2004). "Hypersensitivity to abacavir hypersensitivity conferred by HLA-B*57:01 and a haplotype of HLA-B*57:03-57:01." *Proc Natl Acad Sci U S A* 101(12):4100-5.

Columbo et al. (2006). "The HLA-B*57:01 single-nucleotide polymorphism: a simple screening test for prediction of hypersensitivity reaction to abacavir." *J Infect Dis* 193(2):264-7.

Warfarin (Coumadin®) Sensitivity

Only a medical professional can determine whether warfarin is the right medication for a particular patient. The information contained in this report should not be used to independently establish a warfarin regimen, or abolish or adjust an existing course of treatment.

Warfarin (Coumadin®) Sensitivity

Each time a doctor writes a prescription for warfarin (Coumadin®), a blood thinner given to about two million people each year in the United States, it's a guessing game. There is no "right" dose of the drug. Everyone is different and it can take weeks of adjustment to find a patient's optimal amount of the medication. Too much puts the patient at risk for bleeding. Too little can lead to clots and, in turn, heart attack, stroke or even death. A patient's optimal dose depends not only on age, size, other medications and even diet, but also to a large extent on genetics.

The following results are based on ★★★★★ Established Research for 3 reported markers.

Learn more about the biology of Warfarin Sensitivity.

1 of 3: Warfarin is a drug that can help prevent blood clots.

Your Genetic Data

Who	What it Means	Genes vs. Environment
Jaume Bertranpetit	Substantially increased warfarin sensitivity. May require greatly decreased warfarin dose.	Only a medical professional can determine the right dosage of warfarin for a particular patient. Clinical information such as age, size and other medications the patient is taking can affect a person's optimal dose. The amount of vitamin K in the diet is also a factor. In addition to the genetic variations reported here, there

Warfarin (Coumadin®) Sensitivity

23andMe Established Research Report

Jaume Bertranpetit - March 26, 2012

intended for research and educational purposes. Not for diagnostic use.

Your Genetic Data

Your result - Increased warfarin sensitivity. May require decreased warfarin dose.

Technical Report:

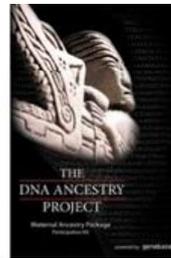
23andMe Name	Genotype	Combination
rs1799853	CT	
rs1057910	AA	CYP2C9 *1/*2, VKORC1 -1639/3673 AA
rs9823231	TT	

Only a medical professional can determine the right medication for a particular patient. This information should not be used to independently establish or adjust an existing regimen.

The case of my individual genome

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Maternal Haplogroup: U5b2b Share

Map History Haplogroup Tree Community

Maternal Haplogroup: U5b2b
U5b2b is a subgroup of U5, which is described below.
Locations of haplogroup U5 circa 500 years ago, before the era of intercontinental travel.

Haplogroup: U5, a subgroup of **I**
Age: 40,000 years
Region: Europe, Near East, North Africa
Populations: Basques, Saami (Sappo), northern Scandinavia
Highlight: Though primarily a European haplogroup, U5 was recently found in mitochondrial DNA extracted from the remains of a 6th-century AD Chinese chieftan.

Your Family and Friends

- [D5a2](#) Japanese Person
- [D5a2a](#) Chinese Person
- [H1e](#) Giovanni Marco Dall'Olio
- [H3](#) Isabel Patricia Fuentes Julia
- [I1](#) Francesc Caballé
- [L3e2b2](#) Nigerian Person
- [U5b2b](#) Jaume Bertrampell

Human Prehistory Videos

Maternal Haplogroup: U5b2b Share

Map History Haplogroup Tree Community

Maternal Haplogroup: U5b2b
U5b2b is a subgroup of U5, which is described below.

Introduction
U5 is one of the oldest haplogroups in Europe. It probably arose when modern humans first moved into western Eurasia from the Near East about 40,000 years ago. As the earliest members of U5 spread across the new territory they would have encountered some oddly familiar inhabitants, the Neanderthals, a close relative of Homo sapiens, that been living in the region for more than 200,000 years. But the Neanderthals proved no match for the new arrivals - by 28,000 years ago they were gone, driven extinct by either competition or outright warfare.

The 6% of Europeans who carry the U5 haplogroup today can trace their maternal ancestry directly back to those early colonizers of Europe. The haplogroup is especially common among the Basque, whose unique language is thought to be descended from that of the first Europeans.

Branches of U5

Haplogroup: U5, a subgroup of **I**
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- [L3e2b2](#) Nigerian Person
- [U5b2b](#) Jaume Bertrampell

Maternal Haplogroup: U5b2b
U5b2b is a subgroup of U5, which is described below.
Click on a region below to highlight the haplogroups common in that region: Sub-Saharan Africa, northern Africa, Near East, Europe, Central Asia, South Asia, Eastern Asia, Oceania, or Americas.

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Age: 40,000 years
Region: Europe, Near East, North Africa
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- [U5b2b](#) Jaume Bertrampell

Famous People

- [H1](#) Mario Anicometti, Luke the Evangelist
- [H3](#) Jimmy Buffet

The case of my individual genome

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Paternal Haplogroup: R1b1b2a1a2c Share

Map History Haplogroup Tree Community

Paternal Haplogroup: R1b1b2a1a2c
R1b1b2a1a2c is a subgroup of R1b1b2, which is described below.
Locations of haplogroup R1b1b2 circa 500 years ago, before the era of intercontinental travel.

Haplogroup: R1b1b2, a subgroup of R1b1

Age: 17,000 years

Region: Europe

Populations: Irish, Basques, British, French

Highlight: R1b1b2 is the most common haplogroup in western Europe, with distinct branches in specific regions.

Your Family and Friends

- [Q2a1b](#) Japanese Person
- [R1b1b2a1](#) Nigerian Person
- [Q2a3b3](#) Frances; Catalan
- [N](#) Chinese Person
- [R1b1b2a1](#) Jaume Bertrampell
- [R1b1b2a1](#) Giovanni Marco Dall'Ole
- [Q2](#) Isabel Patricia Fuentes Julia

Human Prehistory Videos

[Human Prehistory: Prologue](#)

Paternal Haplogroup: R1b1b2a1a2c Share

Map History Haplogroup Tree Community

Paternal Haplogroup: R1b1b2a1a2c
R1b1b2a1a2c is a subgroup of R1b1b2, which is described below.

Introduction
Haplogroup R is a widespread and diverse branch of the Y-chromosome tree that is extremely common in Europe, where it spread after the end of the Ice Age about 12,000 years ago. The haplogroup appears to have originated in southwestern Asia about 30,000 years ago. It then split into two main branches, R1 ultimately spread widely across Eurasia, from Iceland to Japan, whereas R2 mostly remained near its region of origin. Today it can be found in southwestern Asia and India.

Haplogroup R1
R1 is the dominant haplogroup in Europe today, accounting for well over half of all men. After

Haplogroup: R1b1b2, a subgroup of R1b1

Age: 17,000 years

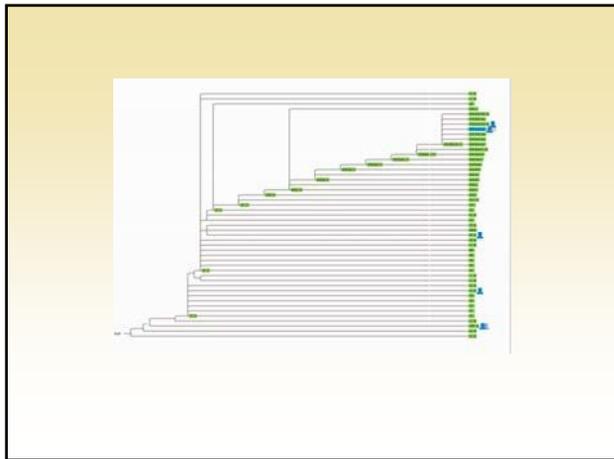
Region: Europe

Populations: Irish, Basques, British, French

Highlight: R1b1b2 is the most common haplogroup in western Europe, with distinct branches in specific regions.

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- [R1b1b2a1](#) Jaume Bertrampell
- [R1b1b2a1](#) Giovanni Marco Dall'Ole
- [Q2](#) Isabel Patricia Fuentes Julia



Paternal Haplogroup: R1b1b2a1a2c
R1b1b2a1a2c is a subgroup of R1b1b2, which is described below.

CKX on a region below to highlight the haplogroups common in that region: Sub-Saharan Africa, Northern Africa, Near East, Europe, Central Asia, South Asia, Eastern Asia, Oceania, or Americas.

Haplogroup: R1b1b2, a subgroup of R1b1

Age: 17,000 years

Region: Europe

Populations: Irish, Basques, British, French

Highlight: R1b1b2 is the most common haplogroup in western Europe, with distinct branches in specific regions.

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Famous People

- [Q1](#) Genghis Khan
- [Q2a1](#) Joseph Stalin
- [Q2a3b1](#) King Louis XVI

- The case of my individual genome
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relative finder

search matches | sort by relationship | 25 per page | << < 1 - 25 of 31 >>

Name	Relationship	Shared DNA	Haplogroups	Actions
Jaume Bertrampell (Male)	You		R1b1b2a1a2c	Update Your Profile
Male	3rd to Distant Cousin	0.20% shared, 1 segment	H1G2, R1b1b2a1a	Send an Introduction
Karen Magnussen-Sonne (Female)	3rd to Distant Cousin	0.19% shared, 1 segment	United States, Christianism; Panama; Ireland; Brooklyn NY, Northern Europe; Magnusen; Sweden, Erickson; 2 more; T2a1	Public Match, Send a Message
Female, b. 1950	3rd to Distant Cousin	0.17% shared, 1 segment	United States; Trinidad/Cuba; Guernsey, Cuba; Hispania/Cuba; Russia; Southern Europe; L3a2b	Send an Introduction
Male	3rd to Distant Cousin	0.17% shared, 1 segment	L3a2b; E1b1b1a2c	Send an Introduction
Male, b. 1970	3rd to Distant Cousin	0.16% shared, 1 segment	United States; USA; E1b1b1c	Send an Introduction
T. William Starr (Male)	3rd to Distant Cousin	0.12% shared, 1 segment	United States; MS; TN; GA; DE; Iowa; Northern Europe; Panama; PRT/Spain; Finland; 21 more; I1a1; R1b1b2a1a1	Introduction Received, Respond
Male	3rd to Distant Cousin	0.12% shared, 1 segment	USA; R1b1b2a1a2c	Send an Introduction

